American Journal of Medical Genetics 70:334–335 (1997)

Letter to the Editor

Reply to Aymé and Philip

To the Editor:

Aymé and Philip [1996] comment on similarities between the patients described in Gripp et al. [1996] and their patient. They conclude that these patients share the same syndrome, as do those in three other previously described cases.

Fine and Lubinsky [1983] described a male with congenital hydrocephalus due to aqueductal stenosis, absence of the corpus callosum, congenital body asymmetry, severe growth failure, and profound developmental delay. Although he shared some findings with our patients (unilateral congenital cataract, small mouth, and inguinal hernia), his overall phenotype and presentation bear little resemblance to those of our patients.

Preus et al. [1984] reported on a male with submucous cleft palate, small posteriorly-rotated ears with a thickened helix and a horizontal top, a small mouth, and unilateral cryptorchidism. His nasal root was described as low and flat, but does not appear low on the lateral photograph (see also Editor's Comment). The nasal tip was wide. His face did not appear flat on lateral view. Sensorineural hearing loss was mentioned in the headline, but not further commented on. The patient had multiple admissions for infections and failure to thrive. Developmental delay was noted, and he had "startle seizures with myoclonic movements." Similarities with our patients include small mouth, inguinal hernia, growth failure, developmental delay, questionable seizures, and possible sensorineural hearing loss. In light of the submucous cleft, ears with thick helix and horizontal top, wide nasal tip, recurrent infections, and growth failure, it would be interesting to study this patient for a microdeletion 22q11.2 (velocardio-facial syndrome; Di George complex).

Suthers et al. [1993] described a male with brachycephaly, deafness, mild cataracts, mental retardation, and hydrocephalus requiring VP-shunt placement. The authors refer to the case of Fine and Lubinsky [1983], but do not consider both patients to share the same syndrome. The patient shared many findings with our patients and was referenced in our report. However, we do not think that he has the same syndrome as our patients, because of differences in the facial findings (e.g., he did not have a broad flare of the alae nasi), because he did not have growth failure, and because our patients did not develop hydrocephalus.

Similarities between our patients and the female seen by Aymé and Philip [1996] are most striking on comparison of her early photographs with our patient 1 as an infant. She later developed a very triangular and asymmetric face, different from that of our patients. Her length is not measured; she is said to be "short". As she can read and write, she appears to be less retarded than our patients. This patients also had similar facial findings to the one described by Suthers et al. [1993]; similarities with the patient reported by Preus et al. [1984] are less striking. The patient in Fine and Lubinsky [1983] is so much more severely affected that it is hard to appreciate the similarities with the case of Aymé and Philip [1996]. Thus, the 4 patients may not have the same syndrome, and "Fine-Lubinsky syndrome" may be a misnomer.

Although the patient described by Aymé and Philip [1996] had many findings in common with our cases, we are not convinced that they share the same syndrome. This issue will probably remain unresolved until a diagnostic test becomes available.

NOTE ADDED IN PROOF

Idiopathic chondrolysis requiring hip replacement had been reported in patient 2 [Gripp et al., 1996]; patient 1 has now been diagnosed similarly. This may be an additional characteristic finding of the underlying syndrome, and will be the subject of a brief clinical report.

REFERENCES

Aymé S, Philip N (1996): Fine-Lubinsky syndrome: A fourth patient with brachycephaly, deafness, cataract, microstomia and mental retardation. Clin Dysmorphol 5:55–60.

*Correspondence to: Linda Nicholson, Division of Medical Genetics, Alfred I. DuPont Institute, Wilmington, DE 19899.

Received 18 July 1996; Accepted 25 July 1996

Fine BE, Lubinsky M (1983): Syndrome identification case report 108: Craniofacial and CNS anomalies with body asymmetry, severe retardation and other malformations. J Clin Dysmorphol 1:6–9.

Gripp KW, Nicholson L, Scott CI Jr (1996): Apparently new syndrome of congenital cataracts, sensorineural deafness, Down syndrome-like facial appearance, short stature, and mental retardation. Am J Med Genet 61:382–386.

Preus M, Cooper AR, O'Leary E (1984): Syndrome identification case report 117: Sensorineural hearing loss, small facial features, submucous cleft palate and myoclonic seizures. J Clin Dysmorphol 1:30–31.

Suthers GK, Earley AE, Huson SM (1993): A distinctive syndrome of brachycephaly, deafness, cataracts and mental retardation. Clin Dysmorphol 2:342–345.

Karen W. Gripp
Charles I. Scott, Jr.
Department of Pediatrics
Thomas Jefferson University
Philadelphia, Pennsylvania

Charles I. Scott, Jr.
Linda Nicholson*
Division of Medical Genetics
Alfred I. DuPont Institute
Wilmington, Delaware